CTSA gene

cathepsin A

Normal Function

The *CTSA* gene provides instructions for making a protein called cathepsin A. Cathepsin A can act as a protease, cutting apart other proteins in order to break them down. Cathepsin A can also act as a protective protein, interacting with other enzymes to prevent them from breaking down prematurely. Based on this protective function, this enzyme is also called protective protein/cathepsin A or PPCA.

Cathepsin A is active in cellular compartments called lysosomes. These compartments contain enzymes that digest and recycle materials when they are no longer needed.

Cathepsin A interacts with the enzymes β-galactosidase and neuraminidase 1, which play a role in the breakdown of complexes of sugar molecules (oligosaccharides) attached to certain proteins (glycoproteins) or fats (glycolipids). Cathepsin A forms a complex with these two enzymes and directs their transport within the cell to the lysosomes. Within lysosomes, cathepsin A activates the enzymes and prevents their breakdown.

On the cell surface, cathepsin A forms a complex with neuraminidase 1 and elastin binding protein, forming the elastin binding protein receptor. This receptor complex plays a role in the formation of elastic fibers, which are a component of the connective tissue that forms the body's supportive framework.

Health Conditions Related to Genetic Changes

galactosialidosis

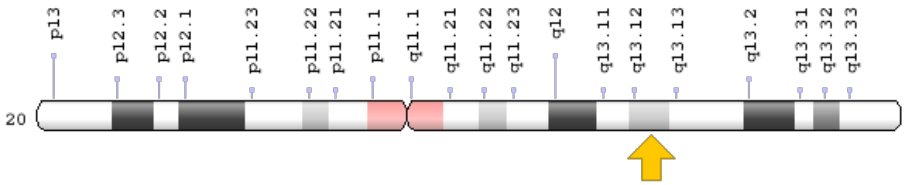
At least 20 mutations in the *CTSA* gene have been found to cause galactosialidosis. Most of these mutations change one protein building block (amino acid) in cathepsin A. In the Japanese population, the most common mutation (written as SpDEx7) disrupts how the gene's instructions are used to make the protein.

Many *CTSA* mutations disrupt the protein structure of cathepsin A, impairing its ability to join with neuraminidase 1 and beta-galactosidase or elastin binding protein. As a result, these other enzymes are not functional or they break down prematurely. Most mutations in the *CTSA* gene cause a lack of functional cathepsin A and a loss of neuraminidase 1, beta-galactosidase, and elastin binding protein. It is not well understood how a lack of these four proteins causes the signs and symptoms of galactosialidosis.

Chromosomal Location

Cytogenetic Location: 20q13.12, which is the long (q) arm of chromosome 20 at position 13.12

Molecular Location: base pairs 45,890,144 to 45,898,820 on chromosome 20 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- beta-galactosidase 2
- beta-galactosidase protective protein
- GSL
- PPCA
- PPGB
- PPGB_HUMAN

Additional Information & Resources

Educational Resources

- Essentials of Glycobiology (second edition, 2009): Lysosomal Degradation of Complex N-Glycans
  https://www.ncbi.nlm.nih.gov/books/NBK1934/#ch41.s3

Scientific Articles on PubMed

- PubMed
  https://www.ncbi.nlm.nih.gov/pubmed?term=%28%28CTSA%5BTIAB%5D%29+OR+%28cathepsin+A%5BTIAB%5D%29%29+OR+%28PPCA%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+2880+days%22%5Bdp%5D

OMIM

- CATHEPSIN A
  http://omim.org/entry/613111

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
  http://atlasgeneticsoncology.org/Genes/GC_CTSA.html
- ClinVar
  https://www.ncbi.nlm.nih.gov/clinvar?term=CTSA%5Bgene%5D
- HGNC Gene Family: Cathepsins
  http://www.genenames.org/cgi-bin/genefamilies/set/470
- HGNC Gene Symbol Report
  http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=9251
- NCBI Gene
  https://www.ncbi.nlm.nih.gov/gene/5476
- UniProt
  http://www.uniprot.org/uniprot/P10619

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